NORTH YORK GENERAL

GENETICS PROGRAM
MOLECULAR GENETICS LABORATORY TESTS

Hematological disorders
Familial Thrombophilia Factor V Leiden and Factor II c.*97G>A
Hereditary Hemochromatosis H63D and C282Y

Infertility
Fragile X-associated Premature Ovarian Insufficiency (FXPOI)
Y-chromosome Microdeletion

Intellectual disability/Autism/Developmental delay
Fragile X Syndrome (FXS)
SNP Array

Myeloid Neoplasms (AML/APL and MDS, MPN, MDS/MPN)
Myeloid Neoplasms NGS Panel (42) - *ABL1, ASXL1, BCOR, BCORL1, BRAF, CALR, CBL, CEBPA, CUX1, CSF3R, DDX41, DNMT3A, EZH2, ETV6, FLT3, IDH1, IDH2, JAK2, GATA2, KIT, KMT2A, KRAS, MPL, NF1, NPM1, NRAS, PHF6, PPM1D, PTPN11, PRPF8, RAD21, RUNX1, SETBP1, SH2B3, SF3B1, SRSF2, STAG,2 TET2, TP53, U2AF1, WT1, ZRSR2*

Neurological disorders
C9orf72-related Disorders
Dentatorubral Pallidoluysian Atrophy (DRPLA)
Fragile X-associated Tremor/Ataxia syndrome (FXTAS)
Friedreich Ataxia (FRDA)
Huntington Disease (HD)
Oculopharyngeal Muscular Dystrophy (OPMD)
Spinal and Bulbar Muscular Atrophy (SBMA)
Spinocerebellar Ataxia (SCA) - SCA1, SCA2, SCA3, SCA6, SCA7, SCA8, and SCA17

Pharmacogenetics
DPYD Genotyping

Prenatal and Neonatal
Aneuploidy of Chromosomes 13, 18, 21, X and Y
SNP Array
Molar Pregnancy

Miscellaneous
Maternal Cell Contamination (MCC)
Specimen Misidentification
Uniparental Disomy of Chromosomes 14 and 15 (UPD14 and UPD15)

Hereditary Cancer
Mutation Analysis - Familial or Tumour findings
Ashkenazi Jewish Founder Panel (9 variants) - *APC* (c.3920T>A), *BRCA1* (c.68_69del; c.5266dup), *BRCA2* (c.5946del), *CHEK2* (c.1283C>T), *GREM1* (40 kb upstream dup), *MSH2* (c.1906G>C), *MSH6* (c.3959_3962del; c.3984_3987dup)
Hereditary Breast/Ovarian/Prostate Cancer Panel (19) - *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
Hereditary Endometrial Cancer Panel (10) - *BRCA1, BRCA2, EPCAM, MLH1, MSH2, MSH6, PMS2, POLD1, POLE, PTEN*
Hereditary GI (Lynch syndrome, Gastric, Pancreas, Polyposis) Cancer Panel (31) - *APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, CTNNA1, EPCAM, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SDHB, SDHD, SMAD4, STK11, TP53*
Hereditary Breast/Ovarian/Prostate/GI Cancer Panel (36) - *APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, CTNNA1, EPCAM*, GALNT12, GREM1, HOXB13 (G84E), MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RNF43, RPS20, SDHB, SDHD, SMAD4, STK11, TP53*
Lynch Syndrome Panel (5) - *EPCAM, MLH1, MSH2, MSH6, PMS2*
Gastric Cancer Panel (17) - *APC, ATM, BRCA1, BRCA2, CDH1, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, SDHB, SDHD, SMAD4, STK11, TP53*
Pancreatic Adenocarcinoma Panel (12) - *ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53*
Polyposis Panel (18) - *APC, BMPR1A, EPCAM, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
Familial Gastrointestinal Stromal Tumor Panel (7) - *KIT, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD*
Familial Melanoma Panel (7) - *BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN*
Familial Renal Cancer Panel (15) - *BAP1, FH, FLCN, MET, MITF, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*
Hereditary Pheochromocytoma and Paraganglioma Panel (12) - *FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
CNS Tumor Panel (20) - *APC, EPCAM, LZTR1, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POLE, POT1, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL*
Soft Tissue Sarcoma Panel (12) - *APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PMS2, TP53*
AXIN2-related Attenuated Familial Adenomatous Polyposis (1) - *AXIN2*
BAP1 Tumour Predisposition Syndrome (1) - *BAP1*
Birt-Hogg-Dube Syndrome (1) - *FLCN*
Carney Complex (1) - *PRKAR1A*
CHRPE, CMV Thyroid, Desmoid (2) - *APC, MUTYH*
DICER-associated Syndrome (1) - *DICER1*

Hereditary Cancer continued
Dysplastic Nevus Syndrome (2) - *CDK4, CDKN2A*
Familial Isolated Pituitary Adenoma (1) - *AIP*
Hereditary Hyperparathyroidism (1) - *CDC73, MEN1*
Hereditary Leiomyomatosis and Renal Cell Cancer (1) - *FH*
Hereditary Lung Cancer - *EGFR*: T790M; V834I; V769M
Li-Fraumeni Syndrome (1) - *TP53*
MEN1 Syndrome (2) - *MEN1, CDKN1B*
Multiple Endocrine Neoplasia Type 2 (1) - *RET*
Neurofibromatosis Type 1 (1) - *NF1*
Gorlin Syndrome (Nevoid Basal Cell Carcinoma Syndrome) (2) - *PTCH1, SUFU*
Nijmegen Breakage Syndrome (1) - *NBN*
Peutz-Jeghers Syndrome (1) - *STK11*
PTEN Hamartoma Tumour Syndrome (1) - *PTEN*
Rare Polyposis Genes (2) - *GALNT12, RPS20*
Retinoblastoma (1) - *RB1*
Rhabdoid Predisposition Syndrome (2) - *SMARCA4, SMARCB1*
Schwannomatosis - *NF2, LZTR1, SMARCB1*
Sessile Serrated Polyposis Cancer Syndrome (1) - *RNF43*
Small Cell Carcinoma of the Ovary, Hypercalcemic Type (1) - *SMARCA4*
Tuberous Sclerosis (2) - *TSC1, TSC2*
Von Hippel-Lindau Syndrome (1) - *VHL*